```
     (FILE 'HOME' ENTERED AT 07:58:53 ON 28 MAY 2003)

     FILE 'MEDLINE, BIOSIS, CAPLUS, EMBASE' ENTERED AT 07:59:04 ON 28 MAY 2003
L1             9 S IZATT-L/AU
               E IZATT-L/AU
               E IZATT-L?/AU
               E IZATT L?/AU
L2            21 S E1-E4
L3            10 DUP REM L2 (11 DUPLICATES REMOVED)

     FILE 'STNGUIDE' ENTERED AT 08:01:16 ON 28 MAY 2003

     FILE 'MEDLINE, BIOSIS, CAPLUS, EMBASE' ENTERED AT 08:04:53 ON 28 MAY 2003
L4         37011 S GENES/SO
L5          8202 S CHROMOSOMES/SO
L6          6521 S L4 AND L5
L7             8 S L6 AND L2
L8             2 DUP REM L7 (6 DUPLICATES REMOVED)
L9             8 S L6 AND IZATT ?/AU
L10            2 DUP REM L9 (6 DUPLICATES REMOVED)
L11           44 S L6 AND ATM
L12           12 DUP REM L11 (32 DUPLICATES REMOVED)

     FILE 'STNGUIDE' ENTERED AT 08:07:54 ON 28 MAY 2003

     FILE 'MEDLINE, CAPLUS' ENTERED AT 08:15:55 ON 28 MAY 2003

     FILE 'STNGUIDE' ENTERED AT 08:15:55 ON 28 MAY 2003

=>
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L Number	Hits	Search Text	DB	Time stamp
1	46	gilad-s$.in. or skaliter-R$.in.	USPAT; US-PGPUB; DERWENT	2003/05/28 07:47
2	38	(gilad-s$.in. or skaliter-R$.in.) and (AT or ATM or breast)	USPAT; US-PGPUB; DERWENT	2003/05/28 07:47
3	7	(gilad-s$.in. or skaliter-R$.in.) and (ATM or breast)	USPAT; US-PGPUB; DERWENT	2003/05/28 07:49
4	8	"5858661"	USPAT; US-PGPUB; DERWENT	2003/05/28 07:52
5	910	telangiectasia and breast	USPAT; US-PGPUB; DERWENT	2003/05/28 07:52
6	837	(telangiectasia and breast) and (mutation$ or alteration or snp or variant)	USPAT; US-PGPUB; DERWENT	2003/05/28 07:55
7	2	"20010021502"	USPAT; US-PGPUB; DERWENT	2003/05/28 07:55

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 s s707p or t2119c or c2119t or p707s
L1             8 S707P OR T2119C OR C2119T OR P707S

=> dup rem l1
PROCESSING COMPLETED FOR L1
L2             6 DUP REM L1 (2 DUPLICATES REMOVED)

=> d ibib ab 1-6

L2   ANSWER 1 OF 6      MEDLINE on STN
ACCESSION NUMBER:    2002719315      MEDLINE
DOCUMENT NUMBER:     22360853    PubMed ID: 12473176
TITLE:               No evidence for association of ataxia-telangiectasia
                     mutated gene T2119C and C3161G amino acid
                     substitution variants with risk of breast cancer.
AUTHOR:              Spurdle Amanda B; Hopper John L; Chen Xiaoqing; McCredie
                     Margaret R E; Giles Graham G; Newman Beth; Chenevix-Trench
                     Georgia; Khanna KumKum
CORPORATE SOURCE:    Cancer and Cell Biology Division, The Queensland Institute
                     of Medical Research, PO Royal Brisbane Hospital,
                     Queensland, Australia.. mandyS@qimr.edu.au
CONTRACT NUMBER:     CA 69638 (NCI)
SOURCE:              Breast Cancer Res, (2002) 4 (6) R15.
                     Journal code: 100927353. ISSN: 1465-5411.
PUB. COUNTRY:        England: United Kingdom
DOCUMENT TYPE:       Journal; Article; (JOURNAL ARTICLE)
LANGUAGE:            English
FILE SEGMENT:        Priority Journals
ENTRY MONTH:         200304
ENTRY DATE:          Entered STN: 20021218
                     Last Updated on STN: 20030403
                     Entered Medline: 20030402
AB   BACKGROUND: There is evidence that certain mutations in the double-strand
     break repair pathway ataxia-telangiectasia mutated gene act in a
     dominant-negative manner to increase the risk of breast cancer. There are
     also some reports to suggest that the amino acid substitution variants
     T2119C Ser707Pro and C3161G Pro1054Arg may be associated with
     breast cancer risk. We investigate the breast cancer risk associated with
     these two nonconservative amino acid substitution variants using a large
     Australian population-based case-control study. METHODS: The
     polymorphisms were genotyped in more than 1300 cases and 600 controls
     using 5' exonuclease assays. Case-control analyses and genotype
     distributions were compared by logistic regression. RESULTS: The 2119C
     variant was rare, occurring at frequencies of 1.4 and 1.3% in cases and
     controls, respectively (P = 0.8). There was no difference in genotype
     distribution between cases and controls (P = 0.8), and the TC genotype was
     not associated with increased risk of breast cancer (adjusted odds ratio =
     1.08, 95% confidence interval = 0.59-1.97, P = 0.8). Similarly, the 3161G
     variant was no more common in cases than in controls (2.9% versus 2.2%, P
     = 0.2), there was no difference in genotype distribution between cases and
     controls (P = 0.1), and the CG genotype was not associated with an
     increased risk of breast cancer (adjusted odds ratio = 1.30, 95%
     confidence interval = 0.85-1.98, P = 0.2). This lack of evidence for an
     association persisted within groups defined by the family history of
     breast cancer or by age. CONCLUSION: The 2119C and 3161G amino acid
     substitution variants are not associated with moderate or high risks of
     breast cancer in Australian women.

L2   ANSWER 2 OF 6  CAPLUS  COPYRIGHT 2003 ACS on STN
ACCESSION NUMBER:        2002:819814  CAPLUS
DOCUMENT NUMBER:         138:104668
TITLE:                   No evidence for association of ataxia-telangiectasia
                         mutated gene T2119C and C3161G amino acid
                         substitution variants with risk of breast cancer
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AUTHOR(S): Spurdle, Amanda B.; Hopper, John L.; Chen, Xiaoqing; McCredie, Margaret R. E.; Giles, Graham G.; Newman, Beth; Chenevix-Trench, Georgia; Khanna, KumKum
CORPORATE SOURCE: Cancer and Cell Biology Division, The Queensland Institute of Medical Research, PO Royal Brisbane Hospital, Queensland, Australia
SOURCE: Breast Cancer Research [online computer file] (2002), 4(6), No pp. given
CODEN: BRCRFS; ISSN: 1465-542X
URL: http://breast-cancer-research.com/content/4/6/R15
PUBLISHER: BioMed Central Ltd.
DOCUMENT TYPE: Journal; (online computer file)
LANGUAGE: English

AB Background: There is evidence that certain mutations in the double-strand break repair pathway ataxia-telangiectasia mutated gene act in a dominant-neg. manner to increase the risk of breast cancer. There are also some reports to suggest that the amino acid substitution variants **T2119C** Ser707Pro and C3161G Pro1054Arg may be assocd. with breast cancer risk. The authors investigated the breast cancer risk assocd. with these two nonconservative amino acid substitution variants using a large Australian population-based case-control study. Methods: The polymorphisms were genotyped in more than 1300 cases and 600 controls using 5' exonuclease assays. Case-control analyses and genotype distributions were compared by logistic regression. Results: The 2119C variant was rare, occurring at frequencies of 1.4 and 1.3% in cases and controls, resp. There was no difference in genotype distribution between cases and controls, and the TC genotype was not assocd. with increased risk of breast cancer (adjusted odds ratio = 1.08, 95% confidence interval = 0.59-1.97). Similarly, the 3161G variant was no more common in cases than in controls (2.9% vs. 2.2%), there was no difference in genotype distribution between cases and controls, and the CG genotype was not assocd. with an increased risk of breast cancer (adjusted odds ratio = 1.30, 95% confidence interval = 0.851.98). This lack of evidence for an assocn. persisted within groups defined by the family history of breast cancer or by age. Conclusion: The 2119C and 3161G amino acid substitution variants are not assocd. with moderate or high risks of breast cancer in Australian women.

REFERENCE COUNT: 13 THERE ARE 13 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L2 ANSWER 3 OF 6 MEDLINE on STN DUPLICATE 1
ACCESSION NUMBER: 2001569957 MEDLINE
DOCUMENT NUMBER: 21518398 PubMed ID: 11606401
TITLE: Spectrum of ATM gene mutations in a hospital-based series of unselected breast cancer patients.
AUTHOR: Dork T; Bendix R; Bremer M; Rades D; Klopper K; Nicke M; Skawran B; Hector A; Yamini P; Steinmann D; Weise S; Stuhrmann M; Karstens J H
CORPORATE SOURCE: Department of Biochemistry and Tumour Biology, Clinic of Obstetrics and Gynecology, Medical School Hannover, D-30659 Hannover, Germany.. thilo.doerk.oststadt@klinikum-hannover.de
SOURCE: CANCER RESEARCH, (2001 Oct 15) 61 (20) 7608-15.
Journal code: 2984705R. ISSN: 0008-5472.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200112
ENTRY DATE: Entered STN: 20011029
Last Updated on STN: 20020122
Entered Medline: 20011204

AB Blood relatives of patients with the inherited disease ataxia telangiectasia (A-T) have an increased susceptibility for breast cancer.

We therefore looked for sequence alterations of the ATM gene in a large hospital-based series of unselected breast cancer patients. The whole ATM coding sequence was analyzed in genomic DNA samples from a core group of 192 consecutive breast cancer cases to define the spectrum of ATM gene mutations. Common sequence alterations were then screened in the whole series of 1000 breast cancer patients and in 500 random individuals. In the core group, 21 distinct sequence alterations were identified throughout the ATM coding region, and 1 common splicing mutation was uncovered in intron 10. Almost half of the breast cancer patients (46%) were heterozygotes for 1 of 16 different amino acid substitutions, and three patients (1.6%) carried a truncating mutation. These data indicate that approximately 1 in 50 German breast cancer patients is heterozygous for an A-T-causing mutation. In our extended series, the most common A-T mutation 1066-6T-->G was disclosed in 7 of 1000 (0.7%) breast cancer patients. Transcript analyses indicated that the loss of exon 11 in the ATM mRNA was the pathogenic consequence of this splicing mutation, which produced a <10% of full-length ATM mRNA and ATM protein in a homozygous A-T patient. We also found an excess of rare missense substitutions in the breast cancer cohort compared with random individuals (7.9% versus 5.3% of alleles; odds ratio = 1.6; P < 0.01). One missense substitution, **S707P** in exon 15, was two times more frequent in breast cancer patients (odds ratio = 2.4; 95% confidence interval, 1.0-5.8) and five times more frequent in patients with bilateral disease than in random individuals (P < 0.001). We conclude that a large variety of distinct ATM mutations and variants exist among breast cancer patients, some of which can contribute to the etiology and progression of the malignancy. Screening for frequent A-T mutations such as the 1066-6-->G splice site substitution can be effective to prospectively identify A-T heterozygotes in an unselected cancer patient population.

L2 ANSWER 4 OF 6 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 2001:53507 BIOSIS
DOCUMENT NUMBER: PREV200100053507
TITLE: Is there a clinical relevance of missense substitutions of the ATM gene in breast cancer patients.
AUTHOR(S): Rades, D. (1); Bremer, M. (1); Kloepper, K.; Doerk, T.; Karstens, J. H. (1)
CORPORATE SOURCE: (1) Radiation Oncology, Hannover Medical University, Hannover Germany
SOURCE: Radiotherapy & Oncology, (September, 2000) Vol. 56, No. Supplement 1, pp. S132. print.
Meeting Info.: 19th Annual Meeting of the European Society for Therapeutic Radiology and Oncology Istanbul, Turkey September 19-23, 2000
ISSN: 0167-8140.
DOCUMENT TYPE: Conference
LANGUAGE: English
SUMMARY LANGUAGE: English

L2 ANSWER 5 OF 6 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 2000:360891 BIOSIS
DOCUMENT NUMBER: PREV200000360891
TITLE: Association of an ATM missense substitution (**S707P**) with breast cancer.
AUTHOR(S): Klopper, Karin (1); Bendix, R. (1); Steinmann, D. (1); Hector, A. (1); Stuhrmann, M. (1); Bremer, M.; Rades, D.; Karstens, J. H.; Doerk, T. (1)
CORPORATE SOURCE: (1) Institute of Human Genetics, Hannover Germany
SOURCE: European Journal of Human Genetics, (June, 2000) Vol. 8 , No. Supplement 1, pp. 109. print.
Meeting Info.: European Human Genetics Conference 2000 Amsterdam, Netherlands May 27-February 30, 2000 European Society of Human Genetics
. ISSN: 1018-4813.

DOCUMENT TYPE: Conference
LANGUAGE: English
SUMMARY LANGUAGE: English

L2 ANSWER 6 OF 6 CAPLUS COPYRIGHT 2003 ACS on STN
ACCESSION NUMBER: 1996:575337 CAPLUS
DOCUMENT NUMBER: 125:218460
TITLE: ATM mutations in cancer families
AUTHOR(S): Vorechovsky, Igor; Luo, Liping; Lindblom, Annika; Negrini, Massimo; Webster, A. David B.; Croce, Carlo M.ennart; Hammarstroem, Lennart
CORPORATE SOURCE: Karolinska Institute, Center BioTechnology, Huddinge, S-14157, Swed.
SOURCE: Cancer Research (1996), 56(18), 4130-4133
CODEN: CNREA8; ISSN: 0008-5472
PUBLISHER: American Association for Cancer Research
DOCUMENT TYPE: Journal
LANGUAGE: English

AB Ataxia-telangiectasia (A-T) is a multisystem recessive disease characterized clin. by cerebellar ataxia, oculocutaneous telangiectasias, immunodeficiency, sensitivity to radiomimetic agents, and cancer predisposition. This pleiotropic disorder is caused by mutations in the ATM (mutated in A-T) gene, which is located in the human chromosomal region 11q22-q23. The ATM gene product is a member of a novel family of large proteins implicated in the regulation of the cell cycle and response to DNA damage. Heterozygosity for A-T was previously suggested to be assocd. with an increased risk of tumors, particularly female breast cancer. Because a loss of constitutional heterozygosity at 11q22-q23 is a frequent event in breast and other tumors, suggesting the presence of a tumor suppressor gene(s) in this region, the authors screened blood DNA samples from 88 unrelated breast cancer patients of Swedish cancer families for ATM mutations using single-strand conformation polymorphism anal. All patients had a family history of tumors previously assocd. with A-T heterozygosity or homozygosity. The authors demonstrate the first three germ-line mutations in ATM identified by screening of breast cancer patients. Two mutations were previously found in A-T homozygotes and one mutation was a 1-bp insertion. All mutations were found in families with a large no. of tumors, however, they did not co-segregate with malignancies. Although the proportion of A-T carriers in this sample seems to be higher than expected by chance, larger studies and pooled data sets will be required to establish that an A-T allele confers cancer susceptibility in heterozygotes.